



C8B gene

complement C8 beta chain

Normal Function

The *C8B* gene provides instructions for making one piece, the beta subunit, of a protein complex called complement component 8. To form this complex, the beta subunit interacts with another piece made up of the alpha subunit (produced from the *C8A* gene) and the gamma subunit (produced from the *C8G* gene), which are linked.

Complement component 8 aids in a part of the body's immune response known as the complement system. The complement system is a group of proteins that work together to destroy foreign invaders, trigger inflammation, and remove debris from cells and tissues. Complement component 8 combines with several other complement proteins to form the membrane attack complex (MAC), which inserts itself in the outer membrane of bacterial cells. This complex creates a hole (pore) in the membrane, which kills the bacterium. This part of the immune response appears to be especially important for fighting infection by bacteria in the *Neisseria* genus.

Health Conditions Related to Genetic Changes

complement component 8 deficiency

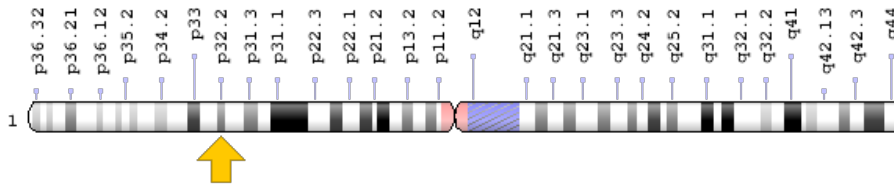
Several mutations in the *C8B* gene cause complement component 8 deficiency type II. This condition is an immune system disorder, known as an immunodeficiency, in which the immune system is not able to protect the body effectively from foreign invaders such as bacteria. People with complement component 8 deficiency have a significantly increased risk of developing recurrent infections, particularly by *Neisseria meningitidis*, which causes meningitis, a serious condition that involves inflammation of the membranes surrounding the brain and spinal cord.

C8B gene mutations involved in complement component 8 deficiency are most often of a type called a C to T transition, in which a DNA building block (nucleotide) called cytosine (C) is changed to the nucleotide thymine (T). Most commonly, this change occurs in a region of the gene called exon 9, but it can occur in other regions. These mutations alter the sequence of protein building blocks, resulting in an abnormally short protein that, if produced, is likely broken down quickly. Other types of mutations can occur in the *C8B* gene; they also lead to an abnormally short protein that is quickly broken down. The resulting shortage of this protein impairs formation of complement component 8. Deficiency of this component prevents formation of membrane attack complexes. Without this part of the immune response, affected individuals are prone to infection, particularly by *Neisseria* bacteria.

Chromosomal Location

Cytogenetic Location: 1p32.2, which is the short (p) arm of chromosome 1 at position 32.2

Molecular Location: base pairs 56,929,210 to 56,974,383 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C82
- complement component 8 subunit beta
- complement component 8, beta polypeptide
- complement component C8 beta chain isoform 1 preproprotein
- complement component C8 beta chain isoform 2
- complement component C8 beta chain isoform 3

Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Terminal Complement Proteins Polymerize to Form Pores in Membranes that can Kill Certain Pathogens
https://www.ncbi.nlm.nih.gov/books/NBK27100/#_A186_
- Molecular Biology of the Cell (fourth edition, 2002): Complement Activation Targets Pathogens for Phagocytosis or Lysis
https://www.ncbi.nlm.nih.gov/books/NBK26846/#_A4679_

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28C8B%5BTIAB%5D%29+OR+%28complement+component+8,+beta+polypeptide%5BTIAB%5D%29%29+OR+%28%28C82%5BTIAB%5D%29+OR+%28complement+component+8+subunit+beta%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+1+preproprotein%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+2%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D>

OMIM

- COMPLEMENT COMPONENT 8, BETA SUBUNIT
<http://omim.org/entry/120960>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=C8B%5Bgene%5D>
- HGNC Gene Family: Complement system
<http://www.genenames.org/cgi-bin/genefamilies/set/492>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1353
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/732>
- UniProt
<http://www.uniprot.org/uniprot/P07358>

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